



PREVENTION OPPORTUNITIES UNDER THE BIG SKY

EXPANDED NEWBORN BLOODSPOT SCREENING IN MONTANA, 2008

Phenylketonuria (PKU) is caused by a block in protein metabolism and congenital hypothyroidism (CH) is due to a deficiency in thyroid hormone production. If not recognized and treated, both cause brain damage soon after birth. More than 40 years ago, state public health laboratories began to screen bloodspots from all newborns for these disorders so early detection and treatment with restricted diet (PKU) or hormone replacement (CH) could save lives and prevent mental retardation. In 2008, Montana joined 44 other states in screening all newborns for the 28 metabolic, endocrine, hematologic, and genetic conditions recommended by the American College of Medical Genetics and the American Academy of Pediatrics.¹ This issue of *Montana Public Health* describes Montana's expanded Newborn (bloodspot) Screening Program.

HOW IT WORKS Newborn screening (NBS) is inexpensive (less than \$100 in laboratory fees) and detects disorders (Table 1) that are individually rare, with incidences from 1 in 3000 to less than 1 in 100,000 births. Tandem mass spectrometry (MS/MS), which was adapted for NBS in the 1990's, quickly quantitates dozens of metabolites in a tiny blood sample to screen for more than 19 conditions. One example is Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD), which occurs in 1 in 12,000 births and blocks fatty acid metabolism. Without NBS, more than 20% of affected babies die before diagnosis, even though treatment is available.² NBS identifies babies at higher risk for diseases so prompt diagnostic testing and treatment can reduce morbidity and mortality. Abnormal results occur in some normal babies (false positives) and some affected infants may not be detected (false negatives). NBS is a public health system that includes: a screening program; follow-up diagnostic testing, and clinical management (including special diets) in cooperation with the baby's medical home; and education.³

Table 1. Required conditions and year mandated, Montana

Year	Conditions Screened
1973	Phenylketonuria (PKU)
1973	Congenital Hypothyroidism (CH)
2003	Galactosemia (GALT)
2003	Hemoglobinopathies (Hb)
2008	Biotinidase Deficiency (BIOT)
2008	Congenital Adrenal Hyperplasia (CAH)
2008	Cystic Fibrosis (CF)
2008	Metabolic Disorders detected by MS/MS
	• Fatty Acid Oxidation (FAO) Disorders (5)
	• Organic Acidemia (OA) Disorders (9)
	• Amino Acid (AA) Disorders (5)

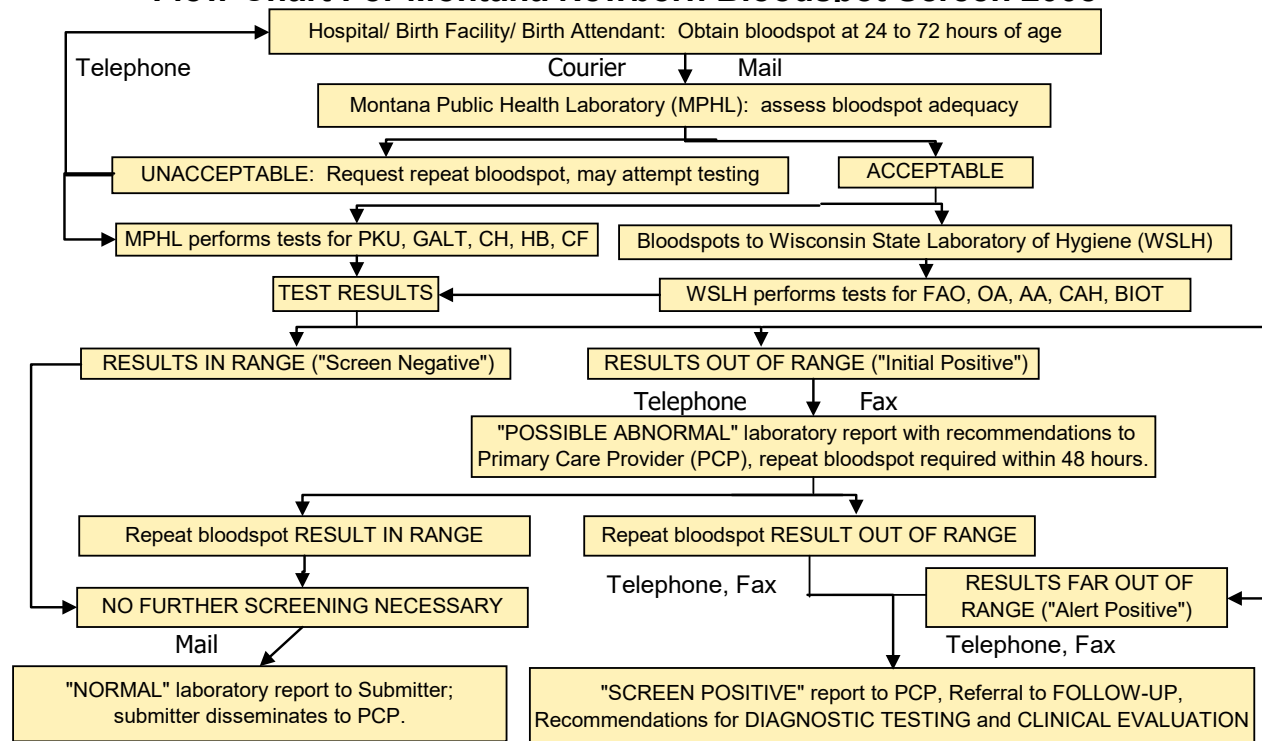
MONTANA'S PROGRAM State law mandated screening for PKU and CH in 1973. Bloodspots were sent to Oregon's state laboratory until 1985, when the Montana Public Health Laboratory (MPHL) began testing. In 2003, screening was mandated for hemoglobinopathies and galactosemia. Since 2002, MPHL has provided the option of screening for additional conditions through the Wisconsin State Laboratory of Hygiene (WSLH). Program monitoring, provision for diagnostic testing, and follow-up for the mandated conditions occurred through Montana Children's Special Health Services (CSHS) specialty clinics.

Table 2. Number of babies screened and test results, 2008

Montana NBS	Jan	Feb	March
Babies screened	1019	966	1027
Initial positive (%)	29 (2.8%)	35 (3.6%)	19 (1.9%)
Screen positive	4	8	5
Confirmed diagnosis	3	5	3

WHAT'S NEW IN 2008 Montana's Administrative Rules⁴ have been amended to require hospitals and birth attendants to obtain bloodspots to screen for 28 conditions between 24 and 72 hours after birth (with an exception for babies with very low birth weights). If any result is abnormal, another bloodspot must be obtained for retesting within 48 hours of that result. If the retest is abnormal, the infant is referred for diagnosis and follow-up (Flow Chart, next page). Screening for the conditions detected by MS/MS is performed by WSLH. The CSHS has contracted with the Montana Medical Genetics Program at Shodair Hospital to coordinate additional diagnostic testing and long-term follow-up associated with expanded NBS. Four babies with congenital hypothyroidism and one with a very rare metabolic disorder were identified in the first three months of expanded screening.

Flow Chart For Montana Newborn Bloodspot Screen 2008



Recommendations for health care providers regarding Newborn Bloodspot Screening

- If you are not sure that NBS was done on your patient in the nursery, collect and submit another bloodspot.
- If notified that a baby has an out-of range result, collect and submit another bloodspot as soon as possible.
- Tell families that Montana NBS now detects more conditions (not just PKU), and this can save babies' lives.

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4. <http://arm.sos.mt.gov/37/37-12555.htm>

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